



IGFBP7 gene

insulin like growth factor binding protein 7

Normal Function

The *IGFBP7* gene provides instructions for making a protein called insulin-like growth factor-binding protein 7 (IGFBP7). Insulin-like growth factors (IGFs) are molecules that are involved in promoting cell growth and division and preventing cells from self-destructing (undergoing apoptosis) prematurely. The IGFBP7 protein is one of a group of proteins that help control the availability of IGFs in body fluids and tissues and increase or decrease the attachment (binding) of IGFs to other molecules called receptors. The binding of IGFs and their receptors activates the cell signaling processes in which they are involved.

The IGFBP7 protein is active in the lining of blood vessels (the vascular endothelium). Its interactions with IGFs and their receptors are thought to help stop a pathway called BRAF signaling, which is involved in directing cell growth.

Health Conditions Related to Genetic Changes

retinal arterial macroaneurysm with supravulvular pulmonic stenosis

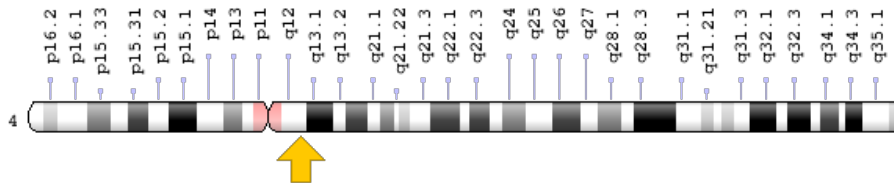
A single *IGFBP7* gene mutation has been identified in people with retinal arterial macroaneurysm with supravulvular pulmonic stenosis (RAMSVPS), a disorder that affects blood vessels in the eyes and heart. In affected individuals, the mutation occurs in both copies of the gene in each cell. The mutation, written as 830-1G>A, is called a splice-site mutation, which means that it affects the way the protein is pieced together. The protein produced from the mutated *IGFBP7* gene is abnormally short and does not function properly.

Without normally functioning IGFBP7 protein to control BRAF signaling, this signaling is increased. It is unknown how this increase is related to the specific blood vessel abnormalities that occur in RAMSVPS, or why these abnormalities are confined to the eyes and the pulmonary artery. Researchers suggest that differences in normal levels of IGFBP7 protein in various parts of the body or the presence of other proteins with a similar function in different tissues may account for the specific signs and symptoms of this disorder.

Chromosomal Location

Cytogenetic Location: 4q12, which is the long (q) arm of chromosome 4 at position 12

Molecular Location: base pairs 57,031,071 to 57,110,385 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AGM
- angiomodulin
- FSTL2
- IBP-7
- IGF-binding protein 7
- IGFBP-7
- IGFBP-7v
- IGFBP-rP1
- IGFBPRP1
- insulin-like growth factor binding protein 7
- MAC25
- PGI2-stimulating factor
- prostacyclin-stimulating factor
- PSF
- RAMSVPS
- TAF
- tumor-derived adhesion factor

Additional Information & Resources

Educational Resources

- Holland-Frei Cancer Medicine (sixth edition, 2003): Classification of Growth Factors and Their Receptors-- The Insulin Family
<https://www.ncbi.nlm.nih.gov/books/NBK12423/#A1066>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IGFBP7%5BTIAB%5D%29+OR+%28insulin-like+growth+factor+binding+protein+7%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- INSULIN-LIKE GROWTH FACTOR-BINDING PROTEIN 7
<http://omim.org/entry/602867>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_IGFBP7.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=IGFBP7%5Bgene%5D>
- HGNC Gene Family: I-set domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/593>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5476
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/3490>
- UniProt
<http://www.uniprot.org/uniprot/Q16270>

Sources for This Summary

- Abu-Safieh L, Abboud EB, Alkuraya H, Shamseldin H, Al-Enzi S, Al-Abdi L, Hashem M, Colak D, Jarallah A, Ahmad H, Bobis S, Nemer G, Bitar F, Alkuraya FS. Mutation of IGFBP7 causes upregulation of BRAF/MEK/ERK pathway and familial retinal arterial macroaneurysms. *Am J Hum Genet.* 2011 Aug 12;89(2):313-9. doi: 10.1016/j.ajhg.2011.07.010.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21835307>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3155176/>
- Dhindsa HS, Abboud EB. Familial retinal arterial macroaneurysms. *Retina.* 2002 Oct;22(5):607-15.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12441727>
- Hooper AT, Shmelkov SV, Gupta S, Milde T, Bambino K, Gillen K, Goetz M, Chavala S, Baljevic M, Murphy AJ, Valenzuela DM, Gale NW, Thurston G, Yancopoulos GD, Vahdat L, Evans T, Rafii S. Angiomodulin is a specific marker of vasculature and regulates vascular endothelial growth factor-A-dependent neoangiogenesis. *Circ Res.* 2009 Jul 17;105(2):201-8. doi: 10.1161/CIRCRESAHA.109.196790. Epub 2009 Jun 18.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19542015>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2936249/>
- OMIM: INSULIN-LIKE GROWTH FACTOR-BINDING PROTEIN 7
<http://omim.org/entry/602867>

Reprinted from Genetics Home Reference:
<https://ghr.nlm.nih.gov/gene/IGFBP7>

Reviewed: August 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services